PCNT gene

pericentrin

Normal Function

The *PCNT* gene provides instructions for making a protein called pericentrin. Within cells, this protein is located in structures called centrosomes. Centrosomes play a role in cell division and the assembly of microtubules. Microtubules are fibers that help cells maintain their shape, assist in the process of cell division, and are essential for the transport of materials within cells.

Pericentrin acts as an anchoring protein, securing proteins to the centrosome that are necessary for its function. Through its interactions with these proteins, pericentrin is involved in the regulation of the cell cycle, which is the cell's way of replicating itself in an organized, step-by-step fashion.

Health Conditions Related to Genetic Changes

microcephalic osteodysplastic primordial dwarfism type II

At least 30 mutations in the *PCNT* gene have been found to cause microcephalic osteodysplastic primordial dwarfism type II (MOPDII). These mutations result in the production of an abnormally short, nonfunctional pericentrin protein that cannot anchor other proteins to the centrosome. As a result, centrosomes cannot properly assemble microtubules, leading to disruption of the cell cycle and cell division. Impaired cell division causes a reduction in cell production, while disruption of the cell cycle can lead to cell death. This overall reduction in the number of cells leads to short bones, microcephaly, and the other signs and symptoms of MOPDII.

prostate cancer

cancers

Gene mutations can be acquired during a person's lifetime and are present only in certain cells. These mutations are called somatic mutations, and they are not inherited. Somatic mutations in the *PCNT* gene can cause an increase in production of the pericentrin protein. Increased levels of pericentrin have been found in solid tumors (including prostate tumors) as well as cancers of blood-forming cells (leukemia and lymphoma). More pericentrin within the centrosome leads to overactivation of the cell cycle and increased cell division. This abnormal cell growth and division can eventually lead to a cancerous tumor.

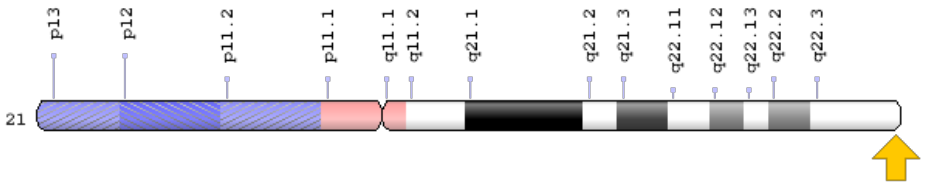
other disorders

Certain common genetic variations (polymorphisms) in the *PCNT* gene have been associated with an increased risk of developing psychiatric disorders such as schizophrenia and depression in some individuals. Similarly, increased levels of pericentrin have been found in some individuals with bipolar disorder. It is unclear how changes in the *PCNT* gene or increased levels of pericentrin protein are related to these disorders. A large number of genetic and environmental factors, most of which remain unknown, likely determine the risk of developing these complex conditions.

Chromosomal Location

Cytogenetic Location: 21q22.3, which is the long (q) arm of chromosome 21 at position 22.3

Molecular Location: base pairs 46,324,103 to 46,445,769 on chromosome 21 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- PCN
- PCNT2
- PCNT_HUMAN
- PCNTB
- pericentrin-2
- pericentrin B

Additional Information & Resources

Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): The Centrosome and Microtubule Organization
  https://www.ncbi.nlm.nih.gov/books/NBK9932/#A1824

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28PCNT%5BTIAB%5D%29+OR+%28pericentrin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- PERICENTRIN
  http://omim.org/entry/605925

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PCNT.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PCNT%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16068
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5116
- UniProt
  http://www.uniprot.org/uniprot/O95613

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